Newborn Screening Quality Assurance Program

Sponsors: Centers for Disease Control and Prevention (CDC) ant the Association of Public Health Laboratories

AN OVERVIEW OF THE PROGRAM

Newborn screening for detection of treatable, inherited metabolic diseases is a major public health responsibility. Effective screening of newborns using dried blood spot (DBS) specimens collected at birth, combined with follow-up diagnostic studies and treatment, helps prevent mental retardation and premature death. These blood specimens are routinely collected from more than 95% of all newborns in the nation; state public health laboratories or their associated laboratories routinely screen DBS specimens for inborn errors of metabolism and other disorders that require intervention. For more than 20 years, the Centers for Disease Control and Prevention (CDC) with their cosponsor the Association of Public Health Laboratories (APHL), has conducted research on materials development and assisted laboratories with the quality assurance (QA) for these DBS screening tests. Most laboratories that test DBS specimens participate voluntarily in our Newborn Screening Quality Assurance Program. Our QA services primarily support newborn screening tests performed by state laboratories; however, CDC also accepts other laboratories and international participants into the QA program.

External quality control (QC) and performance evaluation (PE) programs and materials for DBS screening tests are not available from private-sector organizations. CDC is the sole source of these services for the relatively small number of laboratories testing DBS specimens. CDC provides QC materials, PE services, and technical support to 64 domestic screening laboratories, 20 manufacturer's of diagnostic products, and 134 laboratories in 34 foreign countries.

In 1978, the first DBS materials from CDC were distributed for congenital hypothyroidism screening. Presently, the expanded program includes screening for phenylketonuria, galactosemia, congenital adrenal hyperplasia, homocystinuria, and maple syrup urine disease (MSUD). In 1991, sickle cell disease and other hemoglobinopathies were added to the QA program after studies showed that pneumococcal sepsis in young children with sickle cell disease was reduced by as much as 84% through early identification and treatment; these studies demonstrated the value of newborn screening for this disease. In 1994, the QA program for the DNA confirmatory methods using DBS specimens for sickle cell diseases was added.

The QA program enables screening laboratories to achieve high levels of technical proficiency and continuity that transcends changes in commercial assay reagents while maintaining the high-volume specimen throughput that is required. The PE program provides laboratories with quarterly panels of blind-coded DBS specimens and gives the laboratory an independent external assessment of its performance. During the past year several laboratories misclassified at least one PE specimen and were provided immediate consultation to resolve the analytical problem.

CDC prepares, evaluates and distributes to newborn screening laboratories more than 350,000 DBSs per year. The DBS materials manufactured at CDC must simulate as closely as possible the actual specimens tested by the various the assay systems. DBS materials for QC and PE are certified for homogeneity, accuracy, stability, and suitability for all assays manufactured by different commercial sources.

Our interactive efforts with state laboratories continue to strive for improvements in the services offered by the Newborn Screening Quality Assurance Program to meet the growing and changing needs for newborn screening in the public health community.

PROGRAM OPERATIONS

The Newborn Screening Quality Assurance Program (NSQAP) is for laboratories that use dried blood spot specimens to perform newborn screening tests. The purpose of this program is to improve interlaboratory comparability and to work toward interlaboratory standardization. Current participants include newborn screening laboratories, confirmatory testing laboratories that use dried-blood-spot tests, diet monitoring laboratories, and manufacturers.

The QA program has a quality control (QC) part and a performance evaluation (PE) part or proficiency testing (PT). Laboratories can participate in either part or both parts. Results from participating laboratories are identified by laboratory code numbers to assure confidentiality. For the QC part, we distribute dried blood spot samples at 6-month intervals. Participants return quantitative results from five different analytical runs of the QC materials. For each 6-month period, we compile and distribute the reported results. We offer QC programs for thyroxine (T_4), thyroid-stimulating hormone (TSH), phenylalanine (Phe), leucine (Leu), methionine (Met), total galactose (GAL), and 17 α-hydroxyprogesterone (17-OHP).

For the PE part of the program, we distribute quarterly panels of dried blood spot specimens that participants analyze once. They return their analytical results and qualitative (clinical) assessments of the PE specimens. We prepare PE quarterly reports that show the distributions of analytical values and qualitative assessments participants found. We offer PE programs for T_4, TSH, Phe, Leu, Met, GAL, galactose-1-phosphate uridyltransferase, 17-OHP, and biotinidase, and a PE program for sickle cell disease (SCD) and other hemoglobinopathies. (Presently, the PE panels for SCDs and other hemoglobinopathies are limited to samples containing hemoglobins related to SCDs, α-thalassemia, and hemoglobin E-related disorders.)

At the end of each year, we prepare and distribute to all participants a summary of all PE and QC data reported for that year. There is no fee for participation in the NSQAP.

Distributions of PE and QC specimens occur at the beginning of each calendar quarter. Please indicate on the attached form, the analyte(s) that you would like to participate. If you have questions on any part of the QA program, please contact:

Administrative Office
Newborn Screening Quality Assurance Program
Centers for Disease Control and Prevention (CDC)
Chamblee Bldg. 102, Room 2314 (MS-43)
4770 Buford Highway NE
Atlanta, GA 30341-3724
FAX Number: (770) 488-4255

Voice Phone: (770) 488-4093

Newborn Screening Quality Assurance Program

PARTICIPATION REQUEST FORM

This laboratory would like to enroll in the indicated parts of the Newborn Screening Quality Assurance Program for dried-blood-spot tests. Please place a check mark on the appropriate line(s).

Performance Evaluation	ENROLL
Inborn Errors of Metabolism	_____
Sickle Cell Disease/Other Hemoglobinopathies	_____
Biotinidase Deficiency	_____
Galactose-1-Phosphate Uridyltransferase	_____

Quality Control

Thyroxine	_____
Thyroid-stimulating hormone	_____
Phenylalanine	_____
Galactose	_____
17 ⍺ -hydroxyprogesterone	_____
Leucine	_____
Methionine	_____

Requestor's signature: _____

Title: _____

Please send specimens to:

Contact Person: _____

Laboratory Address: _____

_____

_____

Voice Telephone: _____

FAX Number: _____

Please return your completed form to:**Newborn Screening Quality Assurance Program,**

Mailstop F43, Centers for Disease Control and Prevention, 4770 Buford Highway NE, Atlanta, GA 30341-3724 USA. FAX: (770) 488-4255